

Bloom Syndrome

What Your Results Mean

Testing results indicate that you are a carrier of Bloom syndrome. Carriers of Bloom syndrome are not expected to show signs and symptoms of the disease; however, preliminary evidence suggests that carriers may be at a slightly increased risk to develop colorectal cancer. It is recommended that carriers of Bloom syndrome speak to a genetic counselor that specializes in cancer genetics regarding these risks. Risk for current or future pregnancies is dependent on your partner's carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.



It is recommended that you share and discuss this information with all of your health care providers. Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning and their own personal clinical management.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for Bloom syndrome, each of your children has a 1 in 4 (25%) chance to have the condition.

Bloom Syndrome Explained

What is Bloom Syndrome?

Bloom syndrome is an inherited disorder characterized by short stature, skin rashes, and an increased risk to develop cancer. Other features are variable among individuals and may include learning disabilities, sensitivity to the sun, gastroesophageal reflux, recurrent ear and lung infections, and infertility in males. Individuals may also have mild immune system abnormalities that can lead to infections in the ear, lungs, and respiratory tract during infancy.



Prognosis

Prognosis of Bloom syndrome is considered unfavorable. Lifespan is variable but is typically decreased in individuals with a cancer diagnosis. Diagnosis of a tumor is typical around the second decade of life. Individuals with Bloom syndrome usually have normal or near average intelligence.

Treatment

Treatment of affected individuals is symptomatic; there is no current cure for Bloom syndrome. It is usually recommended that individuals with Bloom syndrome avoid sun exposure and undergo increased surveillance for cancer starting in childhood, as cancer is the greatest cause of death in individuals with Bloom syndrome. Some individuals with learning difficulties may also benefit from special education.



Resources

Bloom's Syndrome Association

<http://www.bloomssyndromeassociation.org/>

National Organization for Rare Disorders (NORD)

<https://rarediseases.org/rare-diseases/bloom-syndrome/>

National Society of Genetic Counselors

<https://www.nsgc.org/>